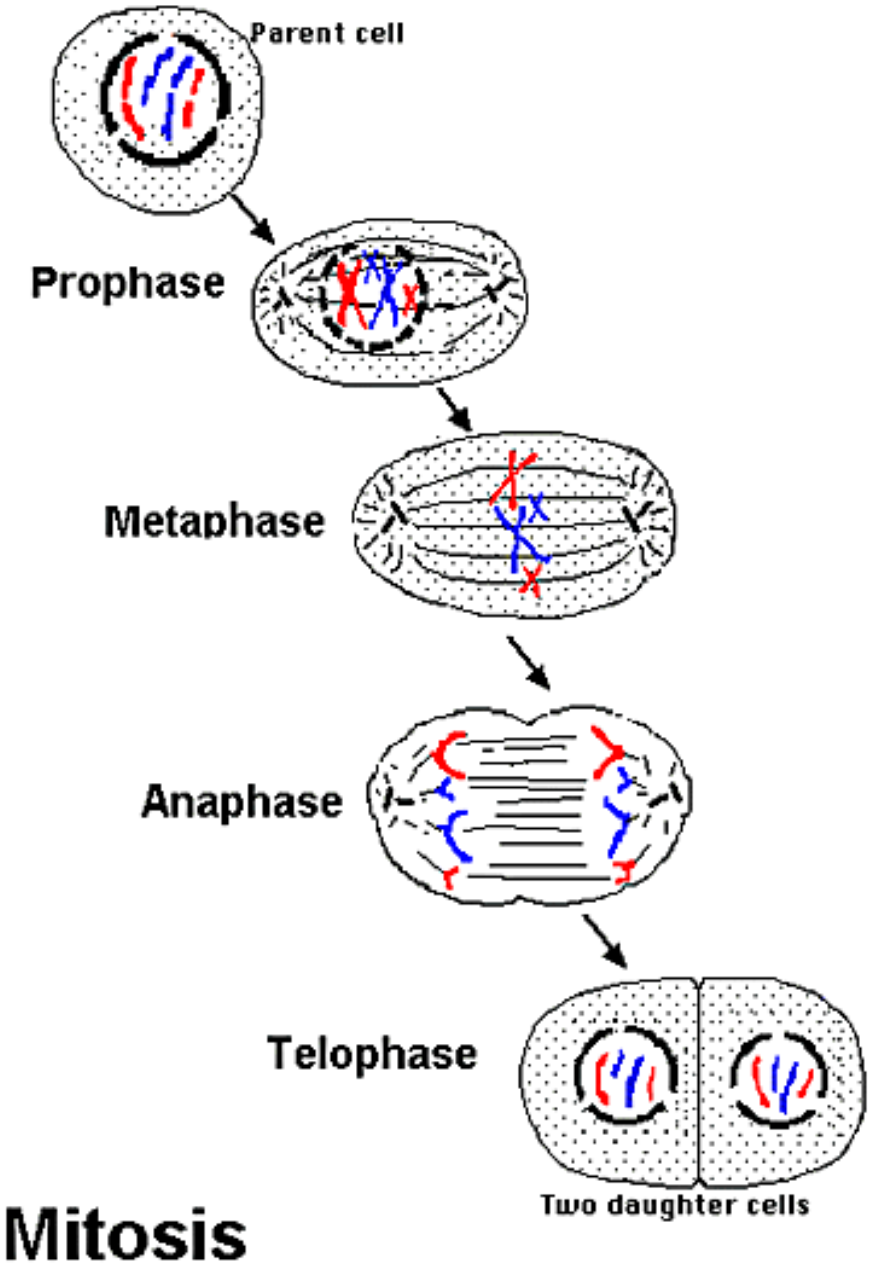
Genetics & Genetic Diseases

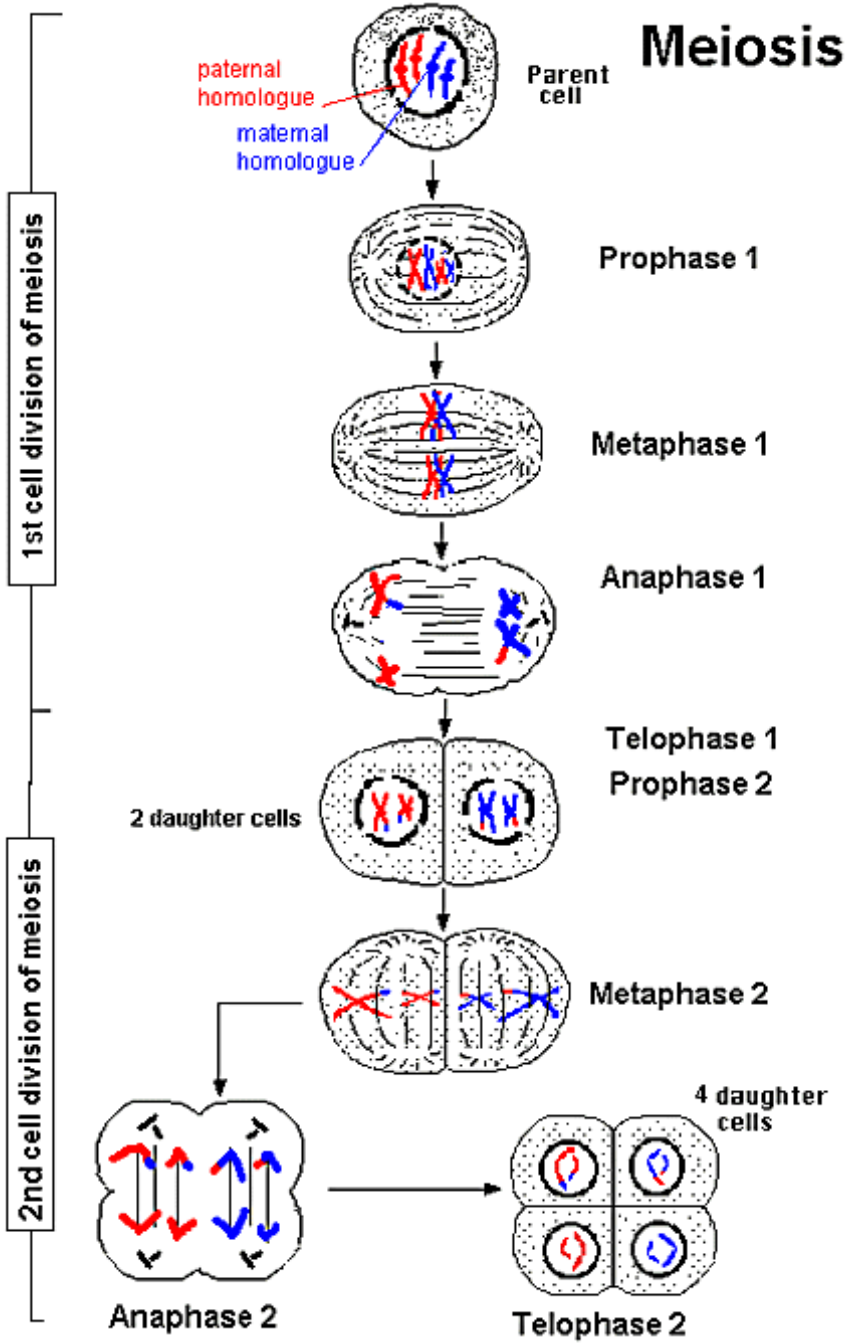
Mendel's Conclusions

- Hereditary characters are determined by discrete factors (genes)
- Genes appear in pairs, one from each parent
- Genes can occur in different forms (alleles)
- If members of gene pair are different (heterozygous), one allele will express itself (dominant allele) and the other will not (recessive allele)

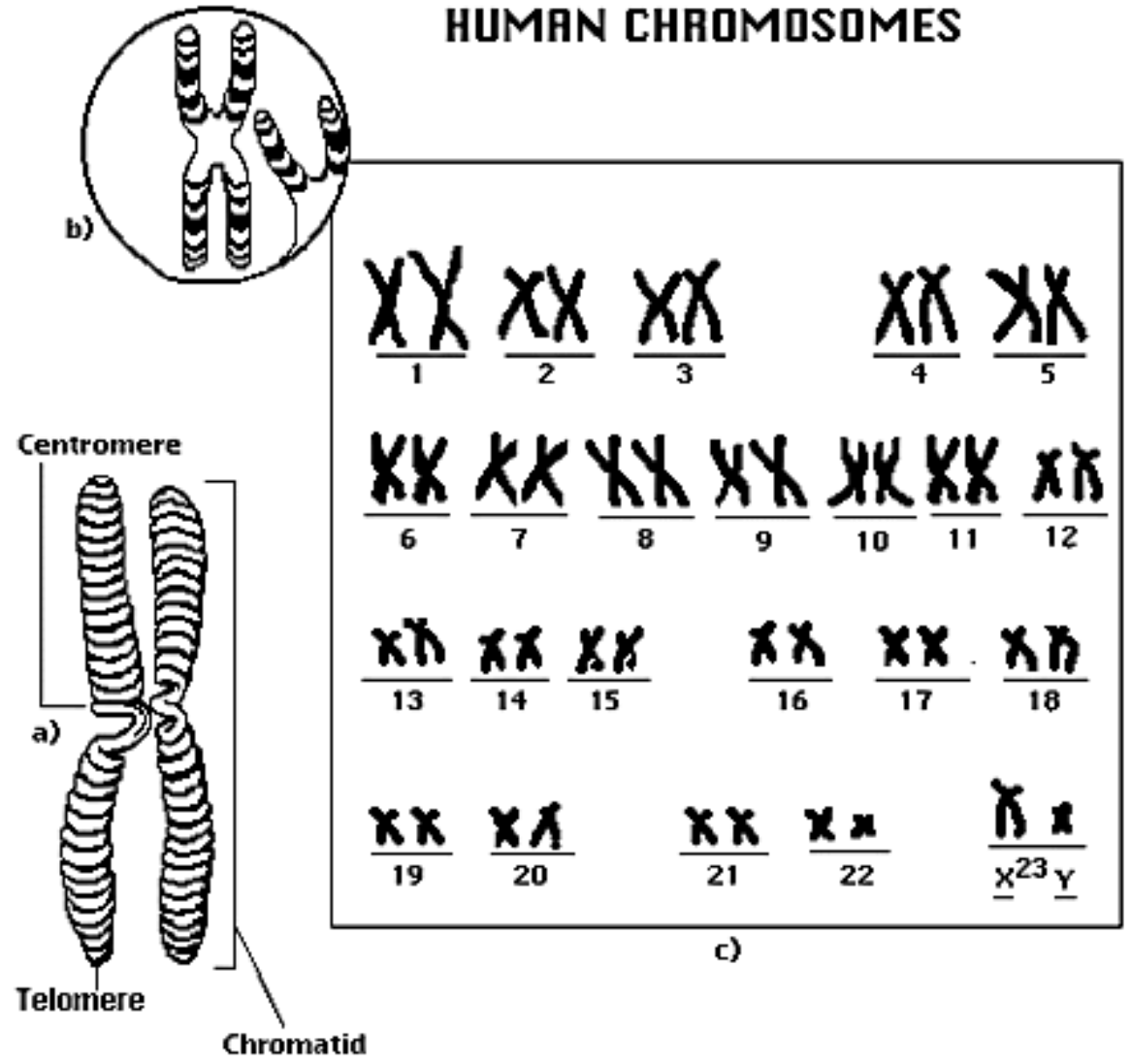
Mitosis


Parent cell
Prophase
Metaphase
Anaphase
Telophase
Two daughter cells
Mitosis

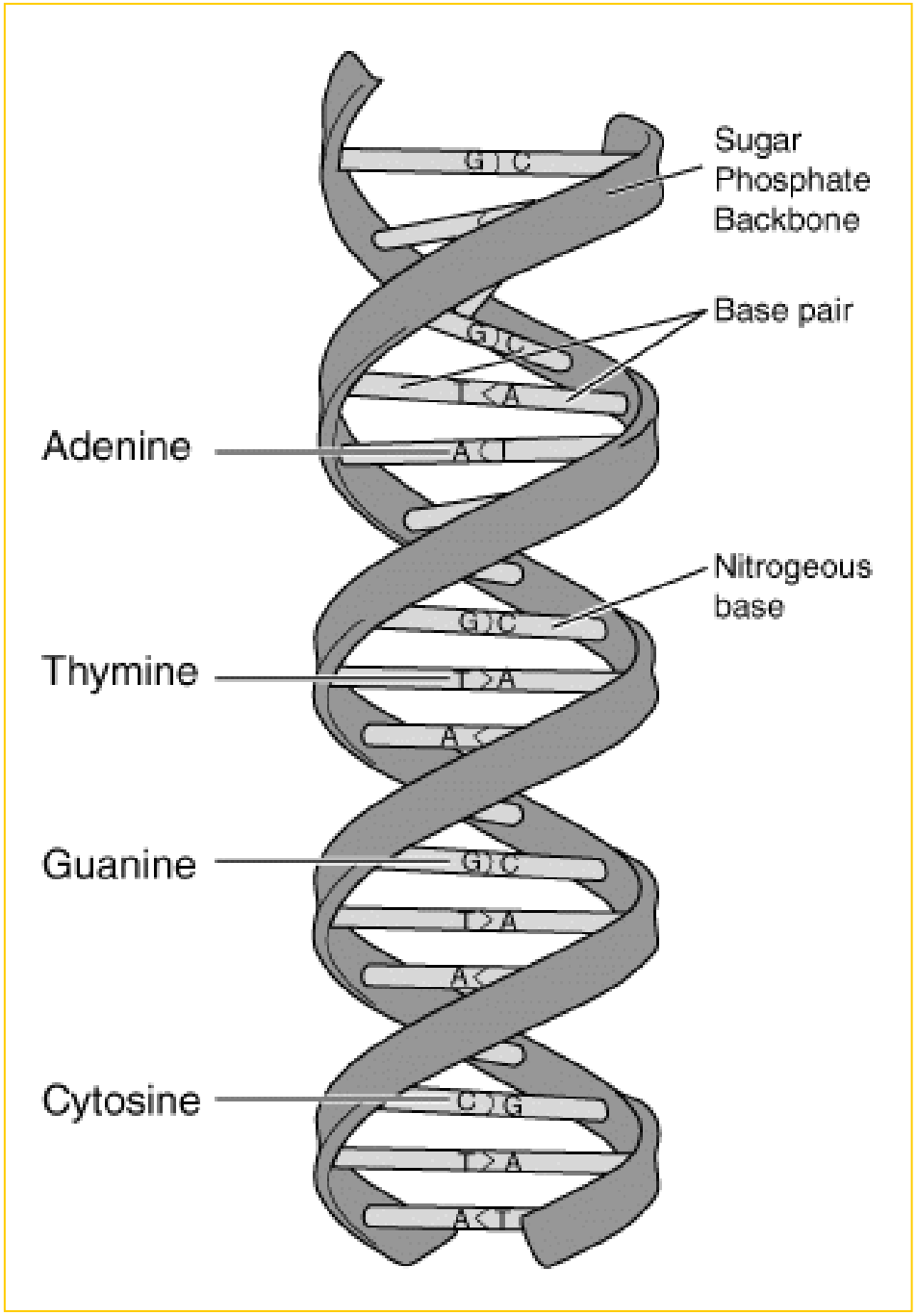
Meiosis


Meiosis
paternal homologue
maternal homologue
Parent cell
Prophase 1
Metaphase 1
Anaphase 1
Telophase 1
Prophase 2
2 daughter cells
Metaphase 2
4 daughter cells
Anaphase 2
Telophase 2
1st cell division of meiosis
2nd cell division of meiosis

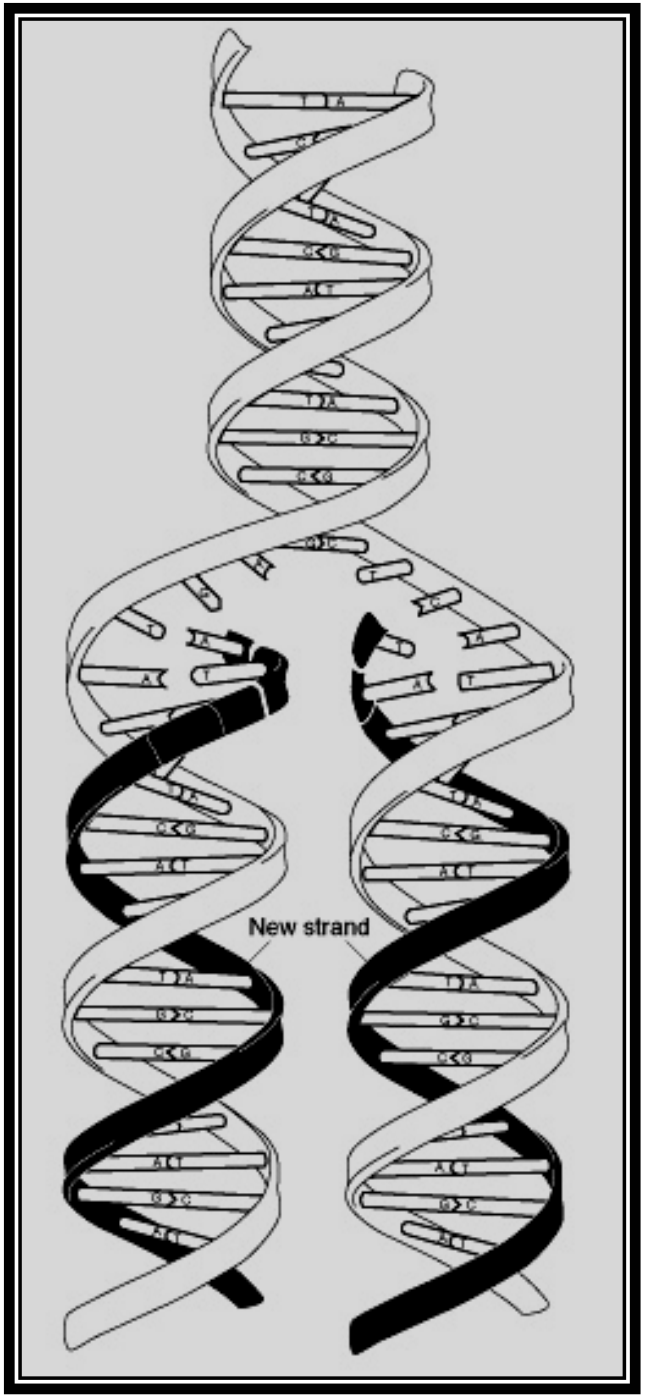
Chromosomes: Carriers of Genes



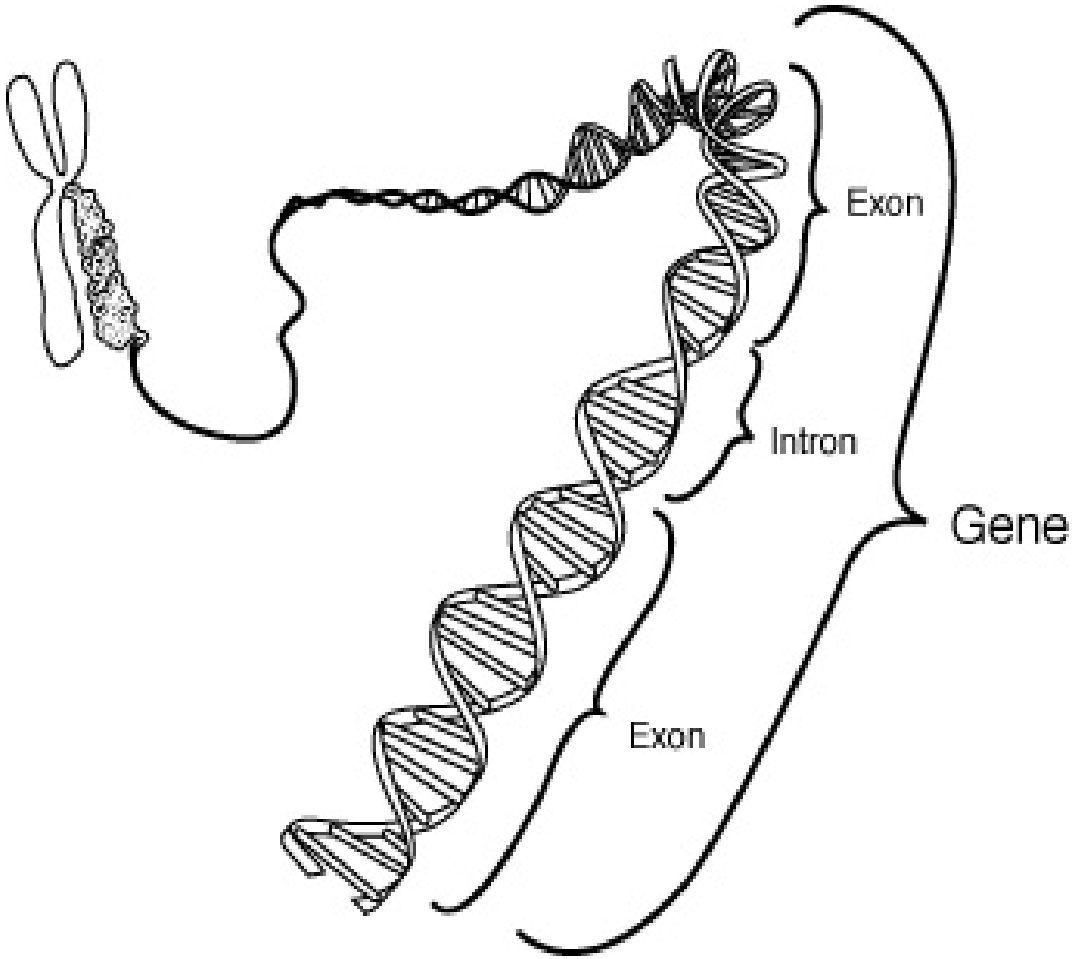
Deoxyribonucleic Acid (DNA)



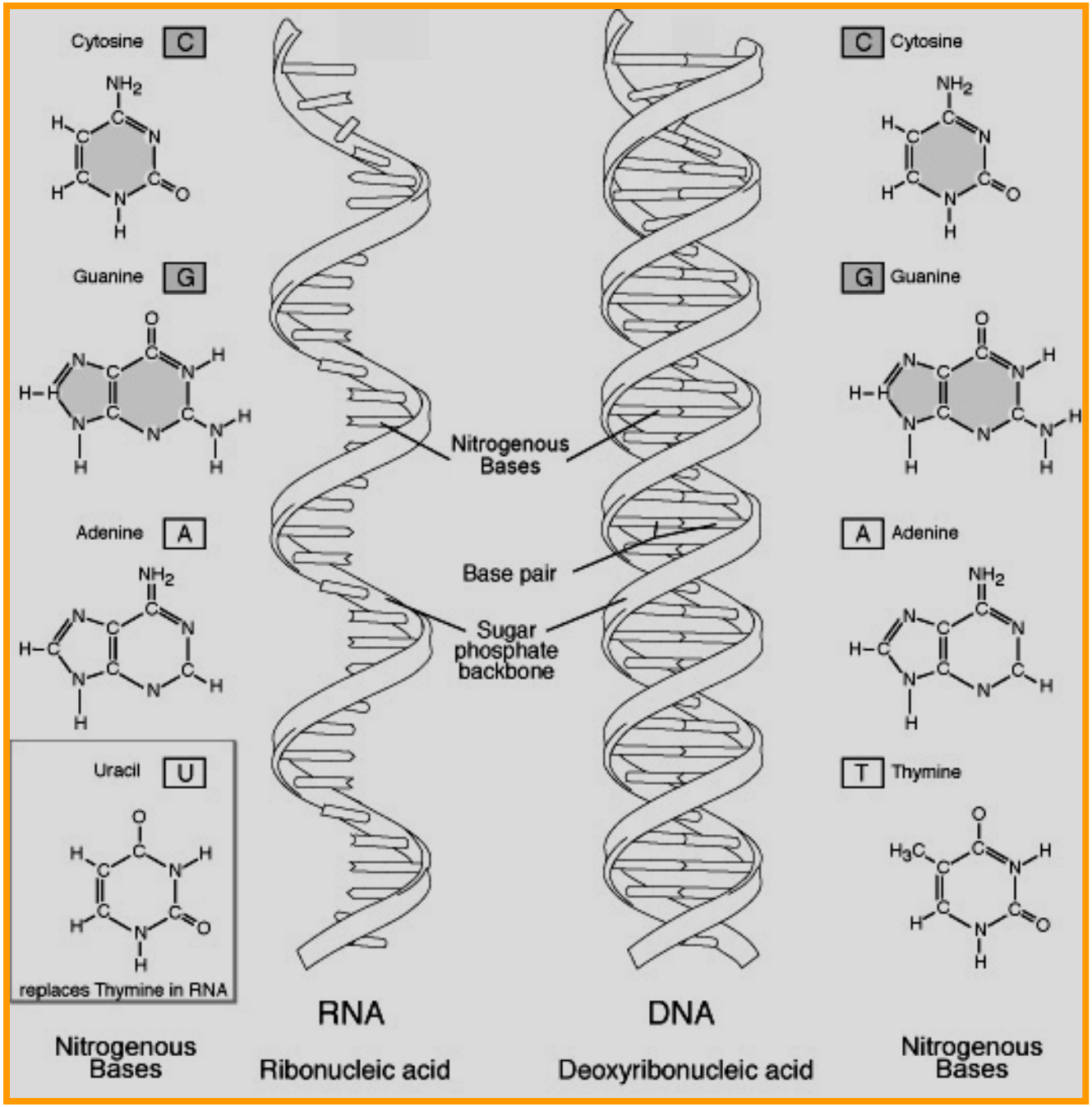
DNA Replication



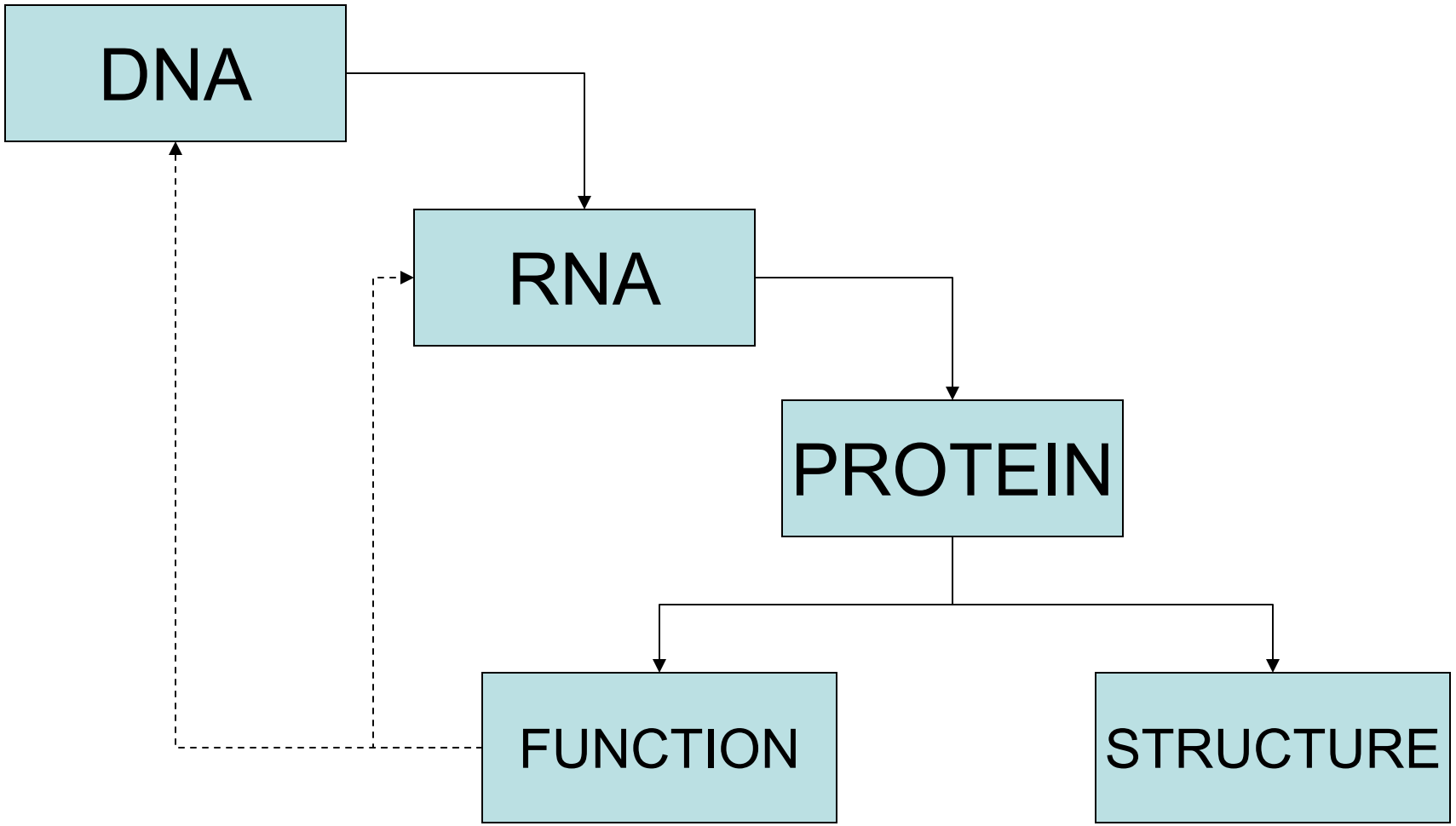
Genes and DNA



Ribonucleic Acid



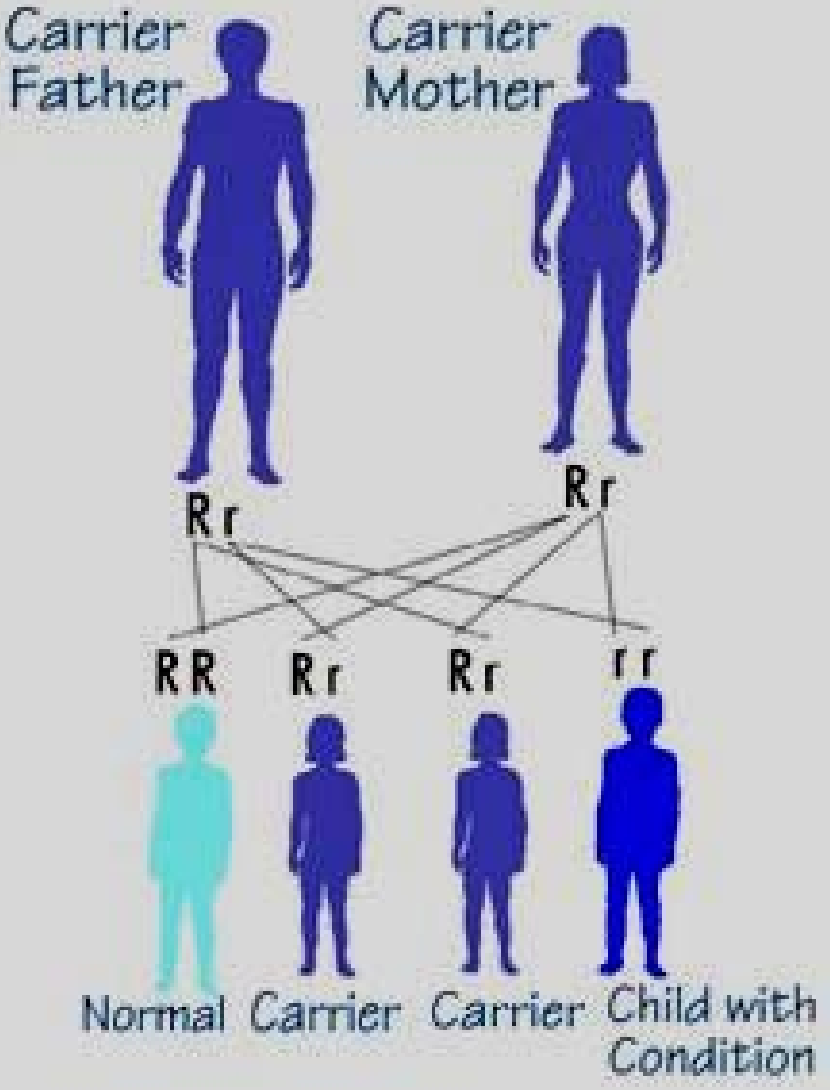
Molecular Biology's Central Dogma



Mendelian Inheritance

- Autosomal Recessive
- Autosomal Dominant
- X-linked Recessive
- X-linked Dominant
- Y-linked

Autosomal Recessive Inheritance



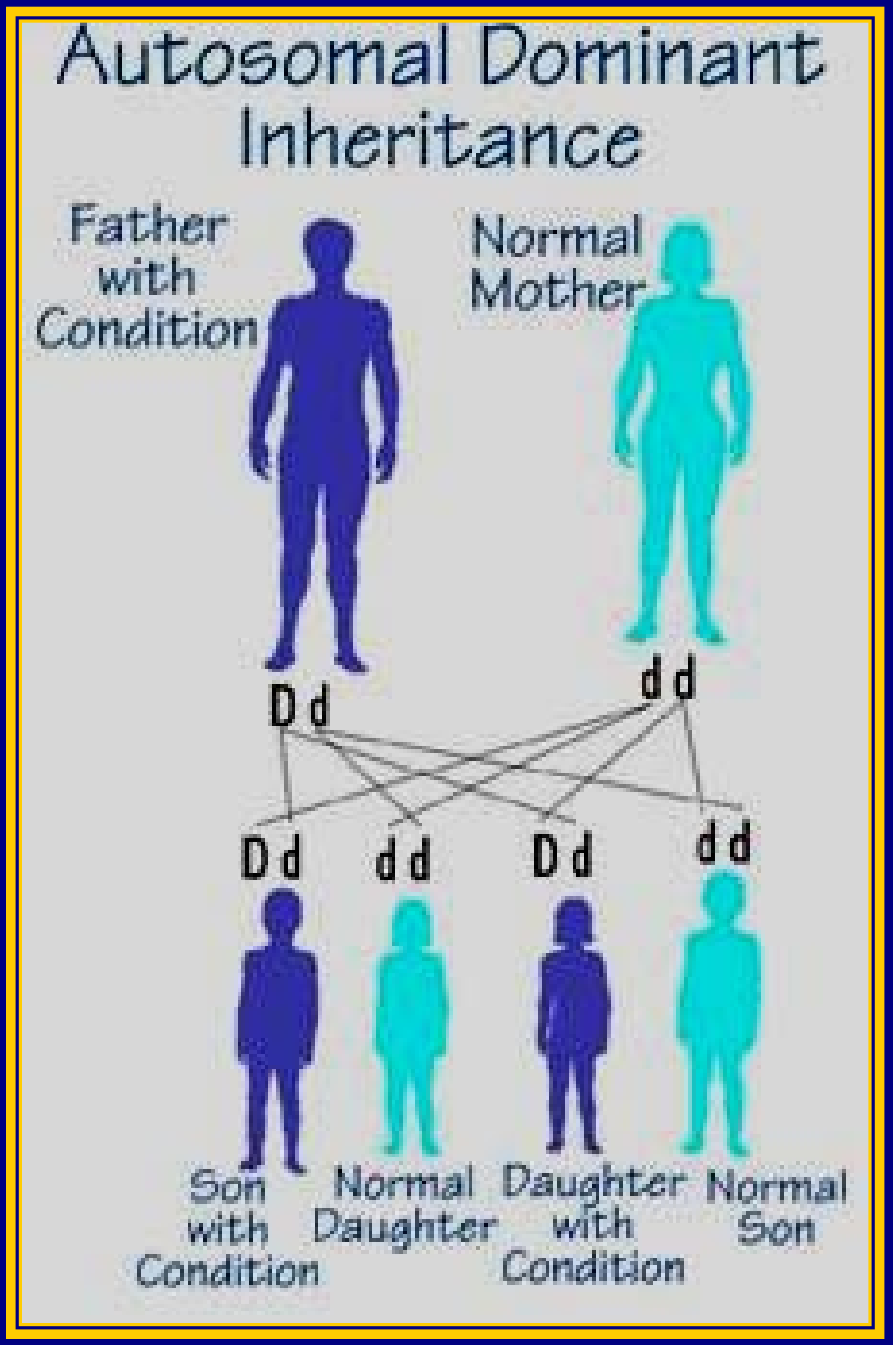
Autosomal Recessive

- Either sex gets disease
- Siblings are usually affected, but not parents
- Heterozygotes of both sexes normal
- Heterozygote parents have diseased kids in ratio of 1:3
- If defect involves a rare gene parents may be related

Autosomal Recessive

- Galactosemia
- Tay-Sachs Disease
- Cystic Fibrosis
- Albinism
- Phenylketonuria (PKU)
- Sickle Cell Anemia

Autosomal Dominant Inheritance



Autosomal Dominant

- Affects males, females equally
- If you have the gene, you have the disease
- Every affected individual has an affected parent
- Normal siblings of affected individuals do not transmit disease (no unaffected “carriers”)

Autosomal Dominant

- Polydactyly
- Achoo Syndrome
- Huntington’s Disease
- Achondroplastic Dwarfism

X-linked Recessive

Father's Gametes

Mother's Gametes

	X^A	Y
X^A	X^AX^A	X^AY
X^a	X^AX^a	X^aY

X-linked Recessive

Father's Gametes

Mother's Gametes

	X^a	Y
X^A	X^AX^a	X^AY
X^A	X^AX^a	X^AY

X-linked Recessive

Father's Gametes

Mother's Gametes

	X^a	Y
X^A	X^AX^a	X^AY
X^a	X^aX^a	X^aY

X-linked Recessive

- More frequent in males
- Passed from affected males through their daughters
- No transmission directly from father to son
- Female carriers can express phenotype at variable levels

X-linked Recessive

- Hemophilia
- Duchene Muscular Dystrophy
- Red/Green Colorblindness

X-linked Dominant

Father's Gametes

Mother's Gametes

	X^A	Y
X^a	X^AX^a	X^aY
X^a	X^AX^a	X^aY

X-linked Dominant

Father's Gametes

Mother's Gametes

	X^a	Y
X^A	$X^A X^a$	$X^A Y$
X^a	$X^a X^a$	$X^a Y$

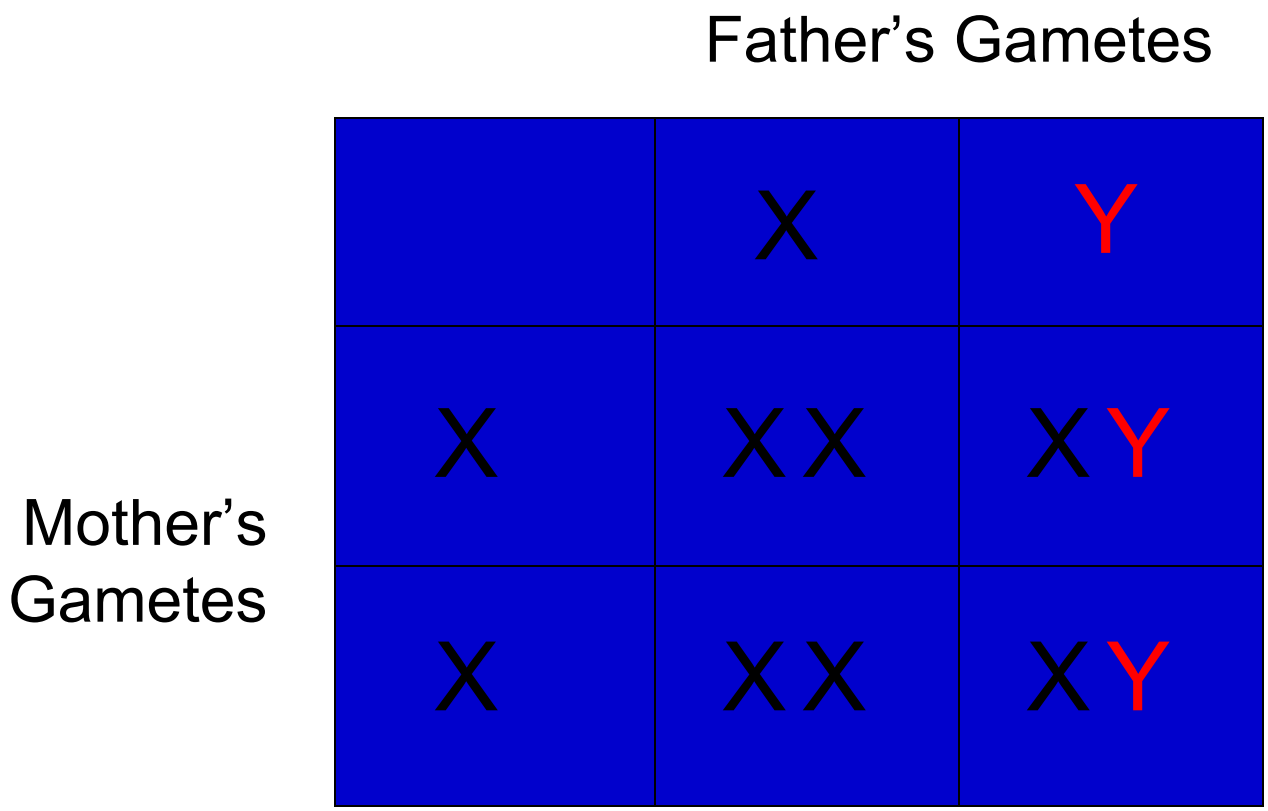
X-linked Dominant

- Affected males with normal spouses have normal sons and all affected daughters
- All offspring have 50% chance of being affected

X-linked Dominant

- Faulty enamel trait

Y-linked



Non-Mendelian Inheritance

- Mitochondrial Traits
  - Mitochondria contain multiple copies of a single DNA strand
  - All mitochondria transmit through ova
  - Mitochondrial traits pass from mother to child
  - Disorders involve combinations of CNS, eye, and muscle tissue abnormalities

Non-Mendelian Inheritance

- Multifactorial Inheritance
  - Do not involve single genes
  - Pedigrees do not exhibit Mendelian patterns
  - Frequently involve interaction between host, environmental factors

Multifactorial Disorders

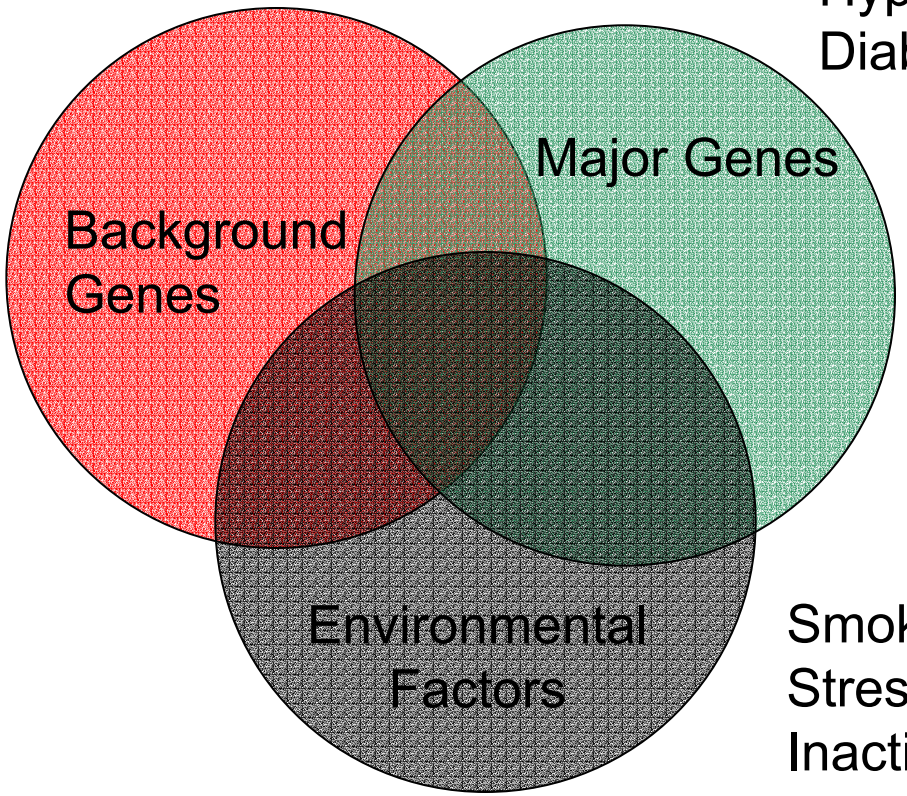
- Congential
  - Neural tube disorders
  - Cleft lip, palette
  - Congenital heart disease

Multifactorial Disorders

- Adult
  - Coronary artery disease
  - Type I diabetes
  - Type II diabetes
  - Breast cancer
  - Colon cancer
  - Lung cancer
  - Rheumatic heart disease
  - Alcoholism
  - Multiple sclerosis
  - Asthma, allergies
  - Autoimmune disorders
  - Bipolar disorder
  - Schizophrenia
  - Kidney stones
  - Gallstones
  - Obesity
  - Peptic ulcer disease
  - Gout

Coronary Artery Disease

Blood pressure
Cholesterol
Triglycerides
HDL
Weight
Glucose



Hypercholesterolemia
Diabetes mellitus

Smoking
Stress
Inactivity
Diet
Oral Contraceptives

Chromosomal Abnormalities

Euploidy

- Addition, loss of complete chromosome sets
- Monoploidy (N)
- Polyploidy
  - Triploidy (3N)
  - Tetraploidy (4N)

Fatal to Embryo

Aneuploidy

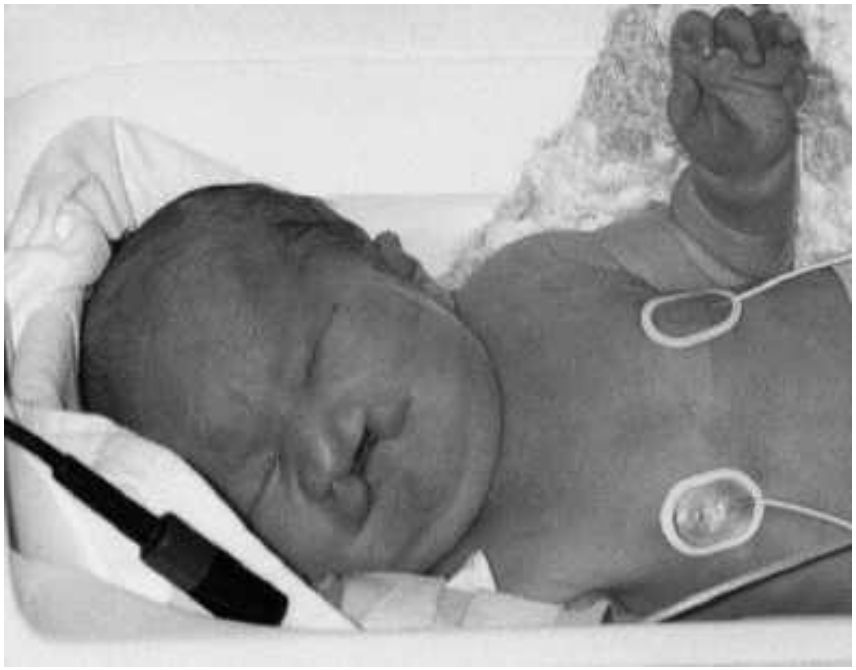
Chromosome abnormalities not involving whole sets

Down's Syndrome

- 1 in 600 births
- Short, broad nose
- Epicanthal fold
- Small oral cavity
- Large, furrowed tongue
- Large, irregular teeth
- IQs from 20 to 50

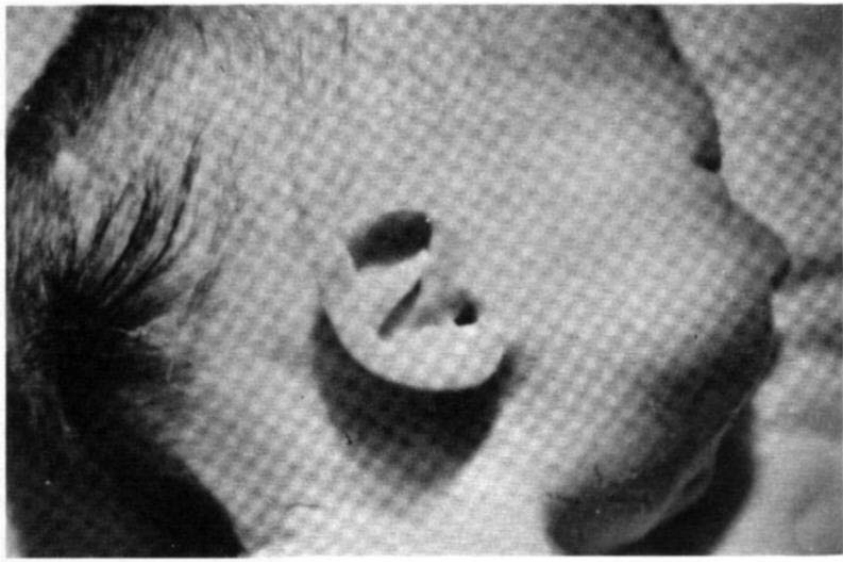
Patau's Syndrome

- 1 in 20,000 births
- Sloping forehead
- Small head, eyes
- Cleft lip, palate
- Heart defects
- 75% die in first year
- 100% by age 6



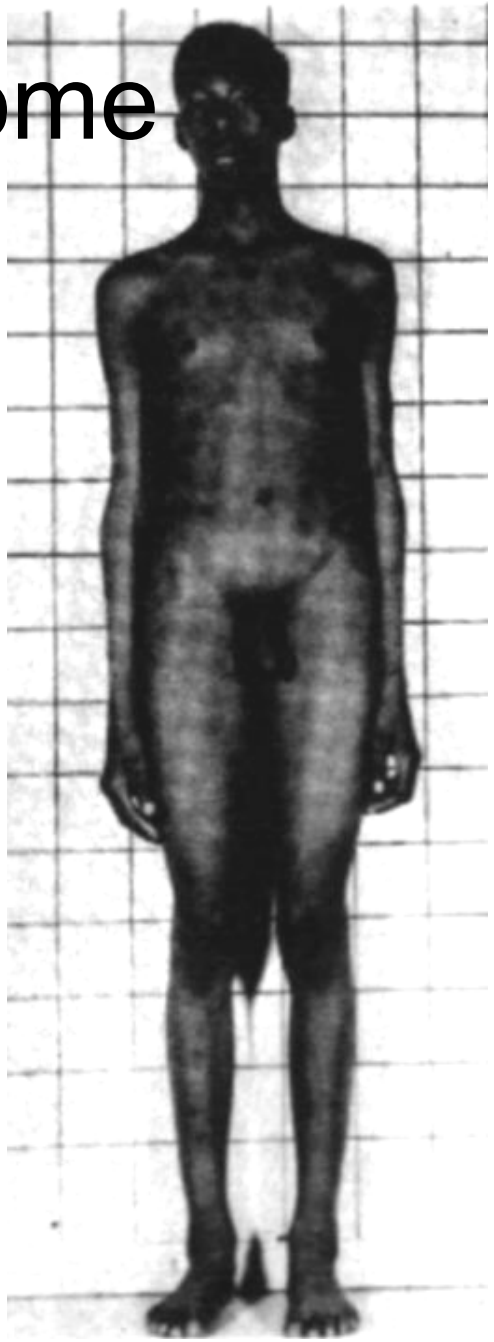
Edwards' Syndrome

- 1 in 11,000 births
- Small mouth, jaw
- Low-set, malformed ears
- Clinched fist, index finger overlapping 3rd, 4th fingers
- Rocker-bottom feet
- Heart defects
- Hearing loss
- 90% die by age 1



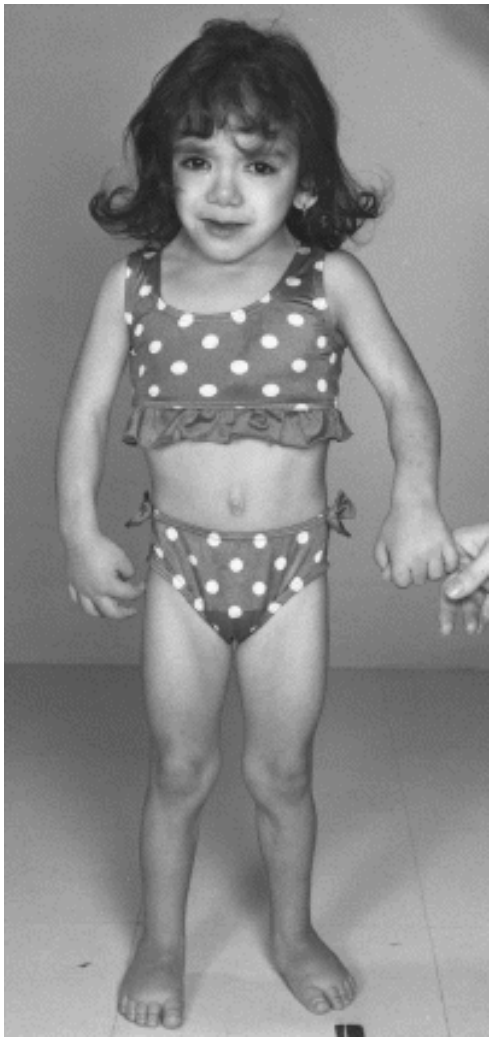
Kleinfelter's Syndrome

- 1 in 500 males
- Taller than average
- Partial breast development
- Small testicles, high-pitched voice, female hair distribution
- Altered body proportions, hips slightly larger than normal
- IQ about 90



Turner's Syndrome

- 1 in 2500 females
- Grow slowly
- Shield chest when young
- Low hair line
- Widely spaced nipples
- Fail to menstruate, no ova
- Normal IQ; weakness in math, spatial perception



Jacob's Syndrome (XYY)

- "Super Male"
- 1 in 1000 males
- 1 in 50 in prison populations
- Excessively tall (2/3s > 6 feet)
- IQs around 80
- Low threshold for control of aggression

X-Polysomy (XXX, XXXX)

- XXX, 1 in 1400 females
- Cannot be distinguished from XX females
- Problems with spontaneous abortions
- XXX have normal IQs, increasing numbers cause mental deficiency